A Searchable Video Database of Dysmorphology

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We have designed a searchable database containing short video clips of patients with a variety of genetic syndromes for the purpose of teaching genetics fellows and counselors to recognize clinical features associated with specific syndromes and to enable research geneticists to place patients with similar dysmorphologies into groups which may represent previously unidentified syndromes.

BACKGROUND AND SIGNIFICANCE

Clinical genetics is an emerging field of medicine which requires the physician to be knowledgeable in diverse areas. Since many genetic alterations have pleiotropic effects, a skilled clinical geneticist can often make an initial diagnosis on the basis of a patient's phenotype. At Baylor College of Medicine, patients seen during the weekly genetics clinic are videotaped to obtain the evaluation of other clinicians and as a teaching tool used to familiarize the fellows and counselors with clinical features of genetic syndromes. The advantage to quickly recognizing a given syndrome is apparent if one considers that a geneticist is often consulted after all other avenues have been exhausted. It is therefore of the utmost importance that the clinical geneticist order the appropriate tests and begin the proper treatment as soon as or even before the presumptive diagnosis has been confirmed. Although a variety of books are available which illustrate classic examples of common syndromes (e.g. Smith's Recognizable Patterns of Human Malformation) and some are even available on CD ROM (London Dysmorphology Index) no one has yet compiled a video catalog of patients during development and representing different ethnic backgrounds for a given syndrome.

Our system allows the physician to search for a specific clinical feature in the clinic note of the patient's visit and then compare the videos of all the patients with this feature who are in the database. In addition, one is able to compare other biochemical (enzyme assays), molecular (DNA diagnostics and Fluorescence In Situ Hybridization [FISH]), physiological (echo cardiograms) and cytogenetic (karyotype) data of these patients simultaneously. In conjunction with HTTP links provided to OMIM and GDB, a physician can compare a recently seen patient to a variety of genetics cases of known diagnosis, review the current literature on the given syndrome and view the expected results of a variety of tests for that disorder.

METHODS

The overall strategy we used was to access the database via the existing HTTP server, MacHTTP, and client Netscape. This resulted in a considerable savings in development effort and allowed us to link through the World Wide Web to On-line Mendelian Inheritance in Man and the Genome Database. By creative use of "Common Gateway Interface" programs and by appropriate localization of large data files, a high performance system has been constructed using low cost hardware.

The local data included in the database consisted of patient clinical records, available as typed hard copy, 1 to 2 minute videotapes of the patients, available as VHS videotapes, karyotypes, available as conventional black and white photographs, and Fluorescent *In Situ* Hybridization (FISH), available as JPEG-encoded computer files. Videotapes were digitized to Quicktime on the built-in hardware of an Apple Macintosh Quadra 840av microcomputer using FusionRecorder software to capture and edit the videos. Karyotypes were digitized on a Apple ColorOne scanner at 300 to 1200 dpi and 256 grey levels. The resulting files were appropriately scaled to fit on the screen and converted to GIF format on a Sun workstation using the "xv" graphics utility. Clinical records were scanned, converted to text with Optical Character Recognition software, and formatted in HTML. The FISH images were linked into the database without modification.

Users of the database are initially given the option of searching by words in the Clinical Records, searching for symptoms contained in a phenotype database we developed, or browsing through a list of syndromes. Once an individual patient is selected the user can view the karyotype, FISH or echo cardiograms. User acceptance of the database has been high.

CONCLUSIONS

1. We have designed a searchable video database to train clinical genetics fellows in recognizing dysmorphology or diagnosing syndromes.
2. We have compiled a variety of information on each patient (Karyotype, FISH etc.) for easy access.
3. We have designed the preliminary system for constructing a database of patients with unknown diagnosis for research purposes.

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